Newborn Screening

Each year, nearly all of the approximately 4 million babies born in the United States are checked for certain medical conditions soon after birth. This is called newborn screening. Newborn screening includes a blood test for several different disorders, hearing screening, and, in some states or hospitals, screening for critical congenital heart defects. Newborns are screened, even if they look healthy, because some medical conditions may not be apparent at birth and may not cause problems until after the first few days or weeks of life. Finding these conditions soon after birth can help prevent some serious problems, such as brain damage, organ damage, and even death.

NCBDDD's Division of Birth Defects and Developmental Disabilities assists states to enhance the quality and usefulness of newborn screening data and programs and ensure that babies receive the full benefits of early identification through newborn screening.

2012 Accomplishments

- Helped states evaluate the effectiveness and costs of newborn screening for critical congenital heart defect (CCHD). CCHD was added to the U.S. Recommended Uniform Screening Panel for newborns in 2011. Evaluating the effect of screening for CCH D will provide states and healthcare providers with data to better understand the possible impact of screening.

- Funded pilot projects in 4 states to collect data on long-term follow-up of children with confirmed newborn screening conditions detected by bloodspot. Follow-up of these children helps guarantee they obtain necessary care and support services for the best possible outcome.

- Worked with other federal agencies and partners organizations on a quality improvement project in primary care pediatric practices to enhance short-term newborn screening follow-up.

Looking to the Future

Newborn screening identifies conditions that can affect a child's health and survival. Long-term follow-up of children with confirmed newborn screening conditions is crucial for maintaining optimal health of these children. Tracking this follow-up is important for public health and is essential for understanding the natural history, prevalence, and management best practices for newborn screening conditions.

For instance, NCBDDD's Division of Birth Defects and Developmental Disabilities is also studying differences between infants identified with certain newborn screening conditions found in states with and without second screening, as well as differences between infants identified on the first screen or the second screen in states with second screening. Routine second testing of newborns for the conditions detected through the newborn screening blood test occurs in 12 states. Opinions differ as to whether routine second testing is the most appropriate public health approach to detect cases that might otherwise be missed by a single newborn screening test. Results from this study will help provide answers for some these question s.

Cora's Story

Cora was born with an undetected congenital heart defect.

To read Cora's story, visit: www.cdc.gov/NCBDDD/PediatricGenetics/stories.html

But it does not end there. NCBDDD's Division of Birth Defects and Developmental Disabilities plans to work closely with programs across CDC and with other agencies to implement, evaluate, and enhance screening programs so that these children have the best possible chance to reach their full potential. For example, we will work with organizations such as the American Academy of Pediatrics to provide continuing medical education courses for pediatric providers on congenital heart defects and newborn screening.

Notable Scientific Publications

CDC. Newborn Screening for Critical Congenital Heart Disease: Potential Roles of Birth Defects Surveillance Programs—United States, 2010-2011. Morbidity and Mortality Weekly Report; 2012; 61: 849-853.

Olney, R. S. and Botto, L. D. Newborn screening for critical congenital heart disease: Essential public health roles for birth defects monitoring programs. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012; 94: 965–969.

Notable Scientific Publications (continued)

Mai, C. T., Riehle-Colarusso, T., O'Halloran, A., Cragan, J. D., Olney, R. S., Lin, A., Feldkamp, M., Botto, L. D., Rickard, R., Anderka, M., Ethen, M., Stanton, C., Ehrhardt, J., Canfield, M. and for the National Birth Defects Prevention Network. Selected birth defects data from population-based birth defects surveillance programs in the United States, 2005–2009: Featuring critical congenital heart defects targeted for pulse oximetry screening. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012; 94: 970–983.

Hinton CF, Neuspiel DR, Gubernick RS, Geleske T, Healy J, Kemper AR, Lloyd-Puryear MA, Saul RA, Thompson BH, Kaye CI. Improving Newborn Screening Follow-up in Pediatric Practices: Quality Improvement Innovation Network. Pediatrics; 2012; 130:e669-75.

Howell RR, Terry S, Tait VF, Olney R, Hinton CF, Grosse S, Eichwald J, Cuthbert C, Popovic T, Glidewell J. CDC Grand Rounds: Newborn Screening and Improved Outcomes. Morbidity and Mortality Weekly Report; 2012; 61:390-393.

To view the annual report online, visit:
www.cdc.gov/ncbddd/2012AnnualReport

For more information about newborn screening, visit:
www.cdc.gov/PediatricGenetics

Did You Know?

- Improvements in technology and endorsement of a uniform newborn-screening panel of diseases have led to earlier life-saving treatment and intervention for thousands of newborns each year.
- About 7,200 babies are born each year in the U.S. with a critical congenital heart defect and are at significant risk of disability or death if their condition is not diagnosed soon after birth.
- Sickle cell disease affects an estimated 100,000 Americans. Finding sickle cell disease early through newborn screening helps to prevent serious health complications as well as deaths from the disease.